The Medical Laboratory Evaluation of a Child Diagnosed With Autism Spectrum Disorder

Confirmation of an autism spectrum disorder (ASD) diagnosis is usually done by a pediatric subspecialist or ideally by a team of ASD or developmental specialists. Although several strategies may be used, all depend on confirmation of Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition, Text Revision, criteria. The Autism Diagnostic Interview-Revised and Autism Diagnostic Observation Schedule are used in most research settings to confirm the presence of these criteria, but expertise in these assessments is often not available in clinical settings. Thus, the specialist(s) must use a battery of evaluation instruments to assess social and language skills and presence of atypical ASD behaviors. Additionally, a thorough history (including health, developmental, behavioral, and family histories), a physical examination, and some evaluation of the child’s overall level of functioning (cognitive, adaptive, and motor skills) are also very important. Finally, one should also assess the family’s resources and coping strategies.

Once the diagnosis of ASD is confirmed by a specialist or team of specialists, the pediatrician plays an important role in the medical evaluation. Although medical investigations include a search for conditions that are known to play an etiologic role in ASD (e.g., fragile X syndrome), they may also be directed at determining the presence of a coexisting medical condition (e.g., seizures, hearing loss). Other factors, such as the availability of local technology and ASD diagnostic teams and/or pediatric subspecialists (developmental pediatricians, neurologists, geneticists, or psychiatrists) with an interest in ASD, will also affect the primary care pediatrician’s role. Some managed care systems may require the primary care pediatrician to order or approve laboratory tests.

An extensive medical laboratory workup is not currently supported by research. This may change as newer technology becomes clinically available and is evaluated. The yield of etiologic evaluation in children without coexisting mental retardation (MR), dysmorphic features, and/or a family history of MR is extremely low. Thus, a thorough laboratory investigation is not recommended in all children with ASDs. However, the clinician must obtain a detailed history (including a 3-generation pedigree) and conduct a thorough physical examination (including a Wood lamp evaluation of the skin) to determine if there are any clinical indications for specific laboratory investigations.

The American Academy of Pediatrics policy statement and accompanying technical report on ASDs suggest a tiered approach for the etiologic workup of a child with ASD.

**Level 1**

An audiologic evaluation should be done in all children with language delay, including those with ASDs. School screening may suffice in older children who can cooperate.

**Level 2**

High-resolution chromosomes (650 bands) and a DNA study for fragile X syndrome should be done in all children with ASDs who have coexisting global developmental delays (GDDs) or, in older children, coexisting MR in addition to ASD. The clinician may want to also consider a fluorescence in situ hybridization (FISH) study to determine the presence of a possible duplication of 15q and/or a methyl CpG binding protein 2 (Rett syndrome) (MECP2) study in females who present with regression.